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110.00 IMPAIRMENTS THAT AFFECT MULTIPLE BODY SYSTEMS

A. What kinds of impairments do we evaluate under this body system listing?

- 1. General. We use these listings when you have a single impairment that affects two or more body systems. Under these listings, we evaluate impairments that affect multiple body systems due to non-mosaic Down syndrome or a catastrophic congenital abnormality or disease. These kinds of impairments generally produce long-term, if not lifelong, interference with age-appropriate activities. Some of them result in early death or interfere very seriously with development. We use the term "very seriously" in these listings to describe an "extreme" limitation of functioning as defined in §416.926a(e)(3).
- 2. What is Down syndrome? Down syndrome is a condition in which there are three copies of chromosome 21 within the cells of the body instead of the normal two copies per cell. The three copies may be separate (trisomy), or one chromosome 21 copy may be attached to a different chromosome (translocation). This extra chromosomal material changes the orderly development of the body and brain. Down syndrome is characterized by a complex of physical characteristics, delayed physical development, and mental retardation. Down syndrome exists in non-mosaic and mosaic forms.
- 3. What is non-mosaic Down syndrome?
- a. Non-mosaic Down syndrome occurs when you have an extra copy of chromosome 21 in every cell of your body. At least 98 percent of people with Down syndrome have this form (which includes either trisomy or translocation type chromosomal abnormalities). Virtually all cases of non-mosaic Down syndrome affect the mental, neurological, and skeletal systems, and they are often accompanied by heart disease, impaired vision, hearing problems, and other conditions.
- b. We evaluate children with confirmed non-mosaic Down syndrome under 110.06. If you have confirmed non-mosaic Down syndrome, we consider you disabled from birth.
- 4. What is mosaic Down syndrome?
- a. Mosaic Down syndrome occurs when you have some cells with the normal two copies of chromosome 21 and some cells with an extra copy of chromosome 21. When this occurs, there is a mixture of two types of cells. Mosaic Down syndrome occurs in only 1-2 percent of people with Down syndrome, and there is a wide range in the level of severity of the impairment. Mosaic Down syndrome can be profound and disabling, but it can also be so slight as to be undetected clinically.
- b. We evaluate children with confirmed mosaic Down syndrome under the listing criteria in any affected body system(s) on an individual case basis, as described in 110.00C.
- 5. What arecatastrophic congenital abnormalities or diseases?
- a. Catastrophic congenital abnormalities or diseases are present at birth, although they may not be apparent immediately. They cause deviation from, or interruption of, the normal

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function of the body and are reasonably certain to result in early death or to interfere very seriously with development.

b. We evaluate catastrophic congenital abnormalities or diseases under 110.08.

B. What documentation do we need to establish that you have an impairment that affects multiple body systems?

- 1. *General*. We need documentation from an acceptable medical source, as defined in §§ 404.1513(a) and 416.913(a), to establish that you have a medically determinable impairment. In general, the documentation should include a clinical description of the diagnostic physical features associated with your multiple body system impairment, and any appropriate laboratory tests.
- 2. Non-mosiac Down syndrome (110.06).
- a. Definitive chromosomal analysis. We will find that you have non-mosaic Down syndrome based on a report from an acceptable medical source that indicates that you have the impairment and that includes the actual laboratory report of definitive chromosomal analysis showing that you have the impairment. Definitive chromosomal analysis for Down syndrome means karyotype analysis. When we have the laboratory report of the actual karyotype analysis, we do not additionally require a clinical description of the physical features of Down syndrome.
- b. What if you have Down syndrome and we do not have the results of definitive chromosomal analysis? When you have Down syndrome and we do not have the actual laboratory report of definitive chromosomal analysis, we need evidence from an acceptable medical source that includes a clinical description of the diagnostic physical features of your impairment, and that is persuasive that a positive diagnosis has been confirmed by definitive chromosomal analysis at some time prior to our evaluation. To be persuasive, the report must state that definitive chromosomal analysis was conducted and that the results confirmed the diagnosis. The report must be consistent with other evidence in your case record; for example, evidence showing your limitations in adaptive functioning or signs of a mental disorder that can be associated with non-mosaic Down syndrome, your educational history, or the results of psychological testing.
- 3. Catastrophic congenital abnormalities or diseases (110.08).
- a. *Genetic disorders*. For genetic multiple body system impairments (other than non-mosaic Down syndrome), such as Trisomy 13 (Patau Syndrome or Trisomy D), Trisomy 18 (Edwards' Syndrome or Trisomy E), chromosomal deletion syndromes (for example, deletion 5p syndrome, also called cri du chat syndrome), or inborn metabolic disorders (for example, Tay-Sachs disease), we need evidence from an acceptable medical source that includes a clinical description of the diagnostic physical features of your impairment, and the report of the definitive laboratory study (for example, genetic analysis or evidence of biochemical abnormalities) that is diagnostic of your impairment. When we do not have the actual laboratory report, we need evidence from an acceptable medical source that is persuasive that

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a positive diagnosis was confirmed by appropriate laboratory analysis at some time prior to our evaluation. To be persuasive, the report must state that the appropriate definitive laboratory study was conducted and that the results confirmed the diagnosis. The report must be consistent with other evidence in your case record.

b. *Other disorders*. For infants born with other kinds of catastrophic congenital abnormalities (for example, anencephaly, cyclopia), we need evidence from an acceptable medical source that includes a clinical description of the diagnostic physical features of the impairment.

C. How do we evaluate impairments that affect multiple body systems and that do not meet the criteria of the listings in this body system?

- 1. These listings are examples of impairments that commonly affect multiple body systems and that we consider significant enough to result in marked and severe functional limitations. If your severe impairment(s) does not meet the criteria of any of these listings, we must also consider whether your impairment(s) meets the criteria of a listing in another body system.
- 2. There are many other impairments that can cause deviation from, or interruption of, the normal function of the body or interfere with development; for example, congenital anomalies, chromosomal disorders, dysmorphic syndromes, metabolic disorders, and perinatal infectious diseases. In these impairments, the degree of deviation or interruption may vary widely from child to child. Therefore, the resulting functional limitations and the progression of those limitations are more variable than with the catastrophic congenital abnormalities and diseases we include in these listings. For this reason, we evaluate the specific effects of these impairments on you under the listing criteria in any affected body system(s) on an individual case basis. Examples of such impairments include, but are not limited to, triple X syndrome (XXX syndrome), fragile X syndrome, phenylketonuria (PKU), caudal regression syndrome, and fetal alcohol syndrome.
- 3. If you have a severe medically determinable impairment(s) that does not meet a listing, we will consider whether your impairment(s) medically equals a listing. If your impairment(s) does not meet or medically equal a listing, we will consider whether it functionally equals the listings. (See §§ 404.1526, 416.926, and 416.926a.) When we decide whether you continue to be disabled, we use the rules in §§ 416. 944a.

110.01 Category of Impairments, Impairments That Affect Multiple Body Systems
110.06 Non-mosaic Down Syndrome, established as described in 110.00B

110.08 A catastrophic congenital adnormality or disease, established as described in 110.00B, and;

A. Death usually is expected within the first months of life, and the rare individuals who survive longer are profoundly impaired (for example, anencephaly, trisomy 13 or 18, cyclopia);

Or

B. That interferes very seriously with development; for example, cri du chat syndrome (deletion 5p syndrome) or Tay-Sachs disease (acute infantile form).